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Cervical Spinal Cord Compression Due to a Plexiform Lesion: A Diagnostic Challenge

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ABSTRACT

Neurofibromatosis is a multisystemic genetic disorder characterized by the development of benign and malignant nervous system tumors, including spinal neurofibromas that can cause significant neurological compromise. Among its rare complications, cervical compressive myelopathy represents a potentially disabling condition that demands early diagnosis and timely surgical intervention. We report the case of a 33-year-old male with a known history of neurofibromatosis who presented with progressive weakness, loss of strength, and spasticity, in whom magnetic resonance imaging revealed multiple cervical neurofibromas causing spinal cord compression from C2 to C4. The patient underwent cervical laminectomy with partial tumor resection, resulting in postoperative neurological improvement. This case underscores the importance of early recognition of neurological symptoms in neurofibromatosis, the essential role of advanced imaging for diagnosis, and the need for a multidisciplinary management approach involving neurology, neurosurgery, rehabilitation, and genetics to optimize functional outcomes and quality of life.

Keywords

Neurofibromatoses, Cervical Myelopathy, Spinal Cord Compression, Spinal Neurofibroma, Laminectomy, Magnetic Resonance Imaging, Central Nervous System.

Introduction

Neurofibromatosis represents a heterogeneous group of genetic disorders characterized by the development of tumors within the nervous system. Among its variants, Neurofibromatosis Type 1 (NF1) is the most prevalent form, accounting for approximately 90% of all neurofibromatosis cases [1]. It is an autosomal dominant disease with an estimated global prevalence of 1 in 3,000 individuals, showing no gender or ethnic predilection. NF1 results from mutations in the NF1 gene, located on chromosome 17, which encodes neurofibromin, a tumor suppressor protein that negatively regulates the RAS signaling pathway, a key regulator of cellular growth. Loss of neurofibromin function leads to uncontrolled cell proliferation and the formation of both benign and malignant tumors [2].

Clinically, NF1 exhibits remarkable phenotypic heterogeneity, with manifestations that include café-au-lait macules, cutaneous and plexiform neurofibromas, axillary or inguinal freckling, Lisch nodules (iris hamartomas), and skeletal abnormalities such as

tibial dysplasia and scoliosis [3]. Systemic complications may involve central and peripheral nervous system tumors, arterial hypertension, cognitive deficits, and skeletal deformities. This wide phenotypic spectrum poses diagnostic and therapeutic challenges, underscoring the importance of multidisciplinary evaluation and long-term follow-up to adequately address the multisystemic nature of the disease [4].

Therapeutic strategies are diverse and may range from surgical resection of symptomatic or deforming tumors to emerging pharmacologic interventions targeting molecular pathways disrupted by NF1 gene mutations [3]. Within the clinical spectrum of NF1, the spinal column is frequently affected [5]. Spinal manifestations may involve both bony structures and soft tissues, often leading to significant neurological complications.

Among the most common spinal manifestations are spinal neurofibromas, benign tumors that may arise at any vertebral level [6]. These lesions can produce pain, weakness, or focal neurological deficits secondary to spinal cord or nerve root compression. Additionally, bony dysplasia and vertebral deformities, such as scoliosis and kyphosis, are frequent findings that can exacerbate neurological or mechanical compromise.

The diagnosis of spinal manifestations in NF1 relies primarily on magnetic resonance imaging (MRI), which provides detailed visualization of tumor extent and spinal involvement [2]. Management depends on the severity and progression of symptoms, ranging from clinical surveillance and conservative management to surgical decompression aimed at relieving spinal cord compression and stabilizing the spine. Surgical interventions are often complex due to the infiltrative nature of neurofibromas and the fragile osseous structures resulting from dysplasia. Consequently, a multidisciplinary approach is essential to optimize therapeutic outcomes and functional recovery.

This case report presents a patient with a confirmed diagnosis of NF1 who developed cervical compressive myelopathy, highlighting the clinical presentation, diagnostic findings, surgical management, and postoperative evolution.

Case Presentation

A 33-year-old male with a history of neurofibromatosis presented with a six-month history of progressive, unintentional weight loss associated with generalized weakness, predominantly affecting the left hemibody. Since childhood, he reported the presence of peripheral tumorous lesions and café-au-lait macules, as well as occasional petechiae. He denied any other relevant personal or family medical history and reported no known allergies.

On physical examination, the patient was alert and oriented to person, time, and place. Pupillary light reflexes were preserved, extraocular movements were intact, facial symmetry was normal, and speech was fluent. Muscle strength was graded 3/5 in the right hemibody and 2/5 in the left hemibody, with marked generalized atrophy and spasticity of the left upper limb. Deep tendon reflexes revealed clonus, positive Hoffmann and Tromner signs, while sensation was preserved. Gait was unsteady due to weakness, and there were no meningeal signs.

Cutaneous examination showed multiple tumorous lesions predominantly in the cervical region, more abundant on the right and occipital sides, as well as café-au-lait spots with irregular but homogeneous borders located in the inguinal, axillary, and cervical regions, accompanied by freckles on the abdomen and thorax.

Routine laboratory tests were within normal limits, with no metabolic abnormalities to explain the patient's clinical presentation. Magnetic resonance imaging (MRI) of the brain, cervical, and thoracic spine revealed multiple enhancing neurofibromas involving the subcutaneous and paravertebral muscle planes bilaterally, as well as spinal neurofibromas along the cervical nerve roots from C1/C2 to T3. These lesions caused neuroforaminal widening and spinal cord compression at the C2–C4 levels, with posterior displacement of the spinal cord, the largest lesion measuring 5 × 1.3 cm in maximum diameter (Figure 1).

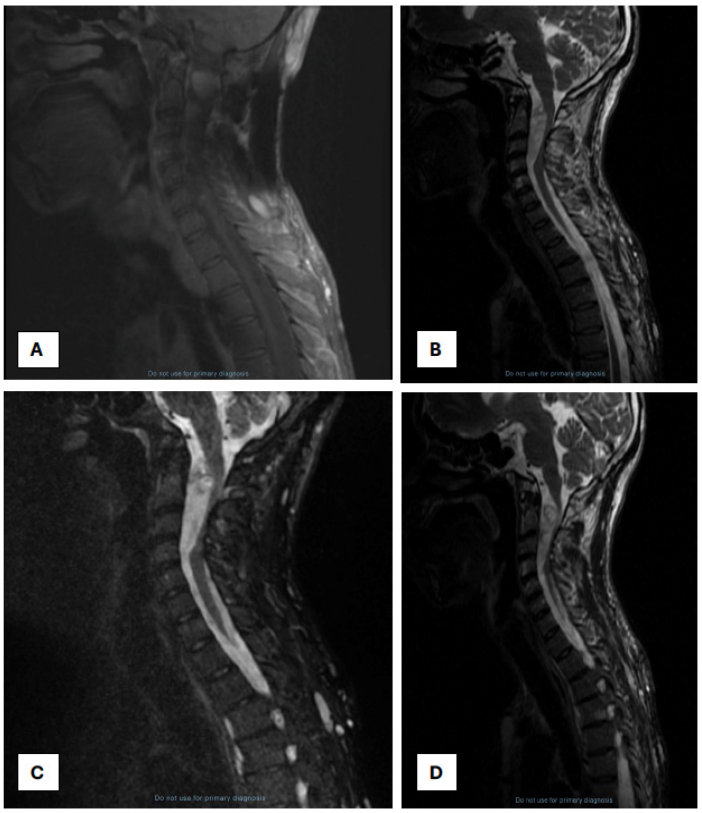

Figure 1: (A) T1-weighted image with contrast showing no enhancement. (B, C, D) STIR sequence: Neurofibromas with contrast enhancement involving the subcutaneous tissue and paravertebral muscle planes bilaterally, as well as spinal neurofibromas along the cervical nerve roots from C1/C2 to T3, causing neuroforaminal widening and spinal cord compression at the C2–C4 levels, with posterior displacement of the spinal cord. The largest lesion measured 5 × 1.3 cm in maximum diameter.

The patient was evaluated by the neurosurgery team, which scheduled a cervical laminectomy, intramedullary tumor resection, dural plasty, and cervical vascular blockade. The procedure was successfully performed without intraoperative complications. During the immediate postoperative period, the initiation of physical therapy was indicated, and follow-up imaging studies were requested (Figure 2).

On physical examination, the patient showed improvement in muscle strength of the left lower limb (3/5), while the left upper limb remained at 2/5, although with a noticeable reduction in spasticity. The patient continued to receive regular physical therapy sessions and remains under clinical follow-up *to assess the long-term response to decompressive surgical treatment.*

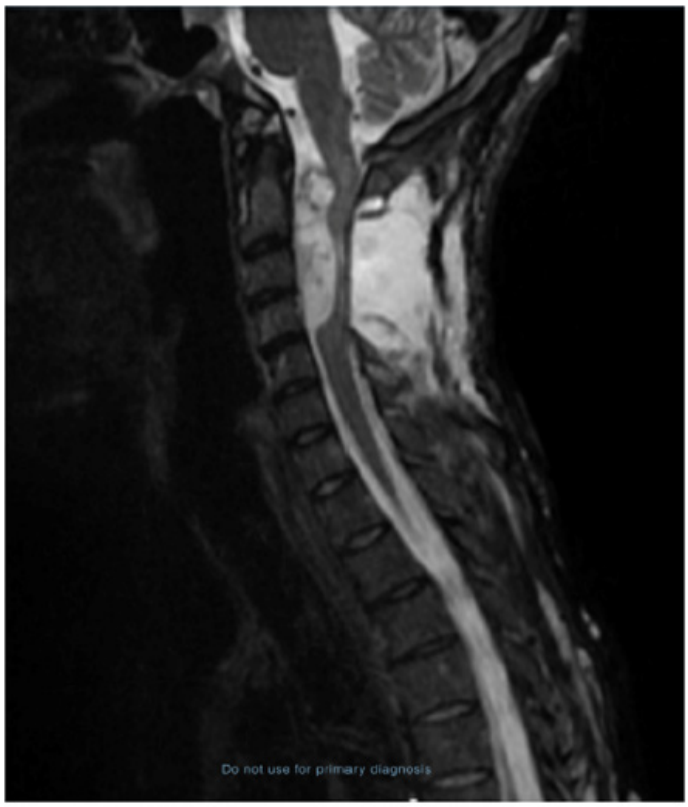

Figure 2: Postoperative changes in the posterior cervical region following decompressive resection of posterior elements from C2 to C4, showing a large fluid collection within the posterior surgical bed, measuring 77 × 25 × 44 mm, exerting a mass effect on the posterior margin of the spinal canal. There are also focal solid lesions consistent with previously known neurofibromas within the spinal canal, located extramedullary in the anterior epidural space, extending from C1 to C4, with expansion of the neuroforamina at C1–C2, C2–C3, C3–C4, and C4–C5. The largest lesion measured 54 × 14 × 16 mm, causing severe spinal cord compression from C2 to C4, with a small hyperintense focus at the C2–C3 level (approximately 8 mm) consistent with myelopathy.

Discussion

Neurofibromatosis is a multisystemic autosomal dominant genopathy with an estimated incidence of approximately 1 in 3,000 live births [1]. It results from loss-of-function mutations in the *NF1* gene, located on chromosome 17q11.2, which encodes neurofibromin, a tumor suppressor protein that negatively regulates the Ras/MAP kinase pathway [4,7,8]. Dysfunction of this protein creates a permissive microenvironment for the development of benign and, occasionally, malignant tumors, predominantly cutaneous, plexiform, and spinal neurofibromas [9].

Spinal involvement has been documented in up to 40% of patients with NF1, with spinal neurofibromas often being multiple and asymptomatic. However, in cases such as the one presented, these lesions may reach a critical volume, resulting in cervical compressive myelopathy [10]. Although uncommon, this complication is associated with progressive functional decline, pyramidal signs, motor weakness, and spasticity, as observed in our patient [11].

Magnetic resonance imaging (MRI) remains the gold standard for diagnosis, as it allows detailed assessment of tumor extent, degree of spinal canal stenosis, and relationship to the spinal cord [12]. In this case, cervical MRI revealed multisegmental spinal neurofibromas with significant compression between C2 and C4, correlating with the patient's neurological findings.

Management of NF1-related myelopathy is primarily surgical in symptomatic cases. Spinal decompression through laminectomy, partial tumor resection, and dural plasty constitute the standard therapeutic approach, although surgical complexity depends on tumor localization, vascularization, and the osseous fragility frequently observed in these patients [13,14]. As illustrated in this case, surgical treatment can achieve partial neurological improvement and halt further clinical deterioration. Postoperative care should include intensive physical rehabilitation to maximize motor and functional recovery [15], along with periodic imaging follow-up to detect possible recurrence or progression of residual lesions [16].

In recent years, targeted pharmacologic therapies have emerged, particularly MEK inhibitors such as selumetinib, which have demonstrated significant benefits in patients with inoperable plexiform neurofibromas, both in pediatric and young adult populations [17,18]. These advances open new avenues for the medical management of NF1, although their use remains limited for symptomatic spinal neurofibromas.

This case emphasizes the importance of early recognition of neurological symptoms in NF1, the appropriate use of imaging techniques, and the timely implementation of surgical interventions. Moreover, it highlights the critical role of a multidisciplinary approach, integrating neurology, neurosurgery, internal medicine, genetics, and rehabilitation, to optimize functional outcomes and enhance the quality of life of affected patients.

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